

Health Care Provider Fact Sheet

Disease Name

Very long-chain acyl-CoA dehydrogenase deficiency

Alternate name(s)
Acronym

N/A
VLCADD

Disease Classification

Fatty Acid Oxidation Disorder

Variants

Yes

Variant name

With and without cardiomyopathy

Symptom onset

Primarily neonatal but some variability.

Symptoms

Hypoketotic hypoglycemia, hepatomegaly, myopathy, cardiomyopathy, adult-onset myopathy.

Natural history without treatment

Sudden infant death due to cardiac abnormalities is common.

Natural history with treatment

Diagnosis and treatment seem to decrease risk for sudden death.

Treatment

Avoidance of fasting, high carbohydrate, low-fat diet supplemented with MCT oil, IV glucose during illness, cornstarch supplementation, avoidance of long chain fatty acids, possible carnitine supplementation.

Other

May have history of a sibling dying of SIDS.

Emergency Medical Treatment

See sheet from American College of Medical Genetics (attached) or for more information, go to website: <http://www.acmg.net/StaticContent/ACT/C14.pdf>

Physical phenotype

No particular dysmorphisms. Cardiomyopathy in infants.

Inheritance

Autosomal recessive

General population incidence

Rare – exact incidence not known

Ethnic differences

None reported

Population

N/A

Ethnic incidence

N/A

Enzyme location

Mitochondrial matrix, heart, liver

Enzyme Function

Long chain fatty acid beta-oxidation

Missing Enzyme

Very long-chain acyl-CoA dehydrogenase

Metabolite changes

Dicarboxylic aciduria, decreased urinary carnitine at times of illness, plasma free carnitine - normal to low, increased plasma long-chain acylcarnitines mildly increased ammonia, lactate and creatine kinase.

Prenatal testing

Enzyme and protein analysis. If a mutation in a proband is detected, DNA prenatal diagnosis via CVS or amniocytes is possible.

MS/MS Profile

Elevated C16:1, C14:2, C14:1, C18:1

OMIM Link

<http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=201475>

Genetests Link

www.genetests.org

Support Group

FOD Family Support Group

<http://www.fodsupport.org>

Save Babies through Screening Foundation

<http://www.savebabies.org>

Genetic Alliance

<http://www.geneticalliance.org>

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